

THE X CHROMOSOME TO THE RESCUE

GENETIC GENEALOGY'S UNSUNG HERO

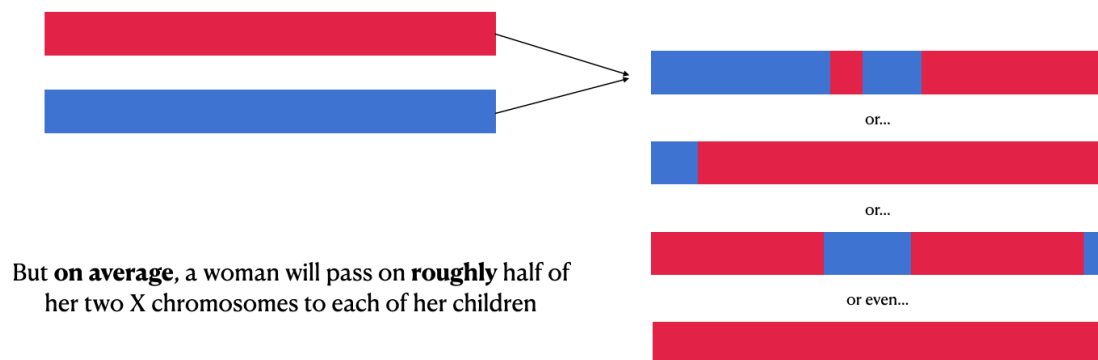
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CHROMOSOME BASICS

- Humans have 23 pairs of chromosomes: 22 pairs of autosomes and 1 pair of sex chromosomes
- 2 types of sex chromosomes are X and Y
- Males typically have XY
- Females typically have XX
- Roughly .0025% of males and .0015% of females have sex chromosome abnormalities such as Turner syndrome (single X in females) or Klinefelter (XXY in males)
- This presentation (and handout) will focus on XY and XX

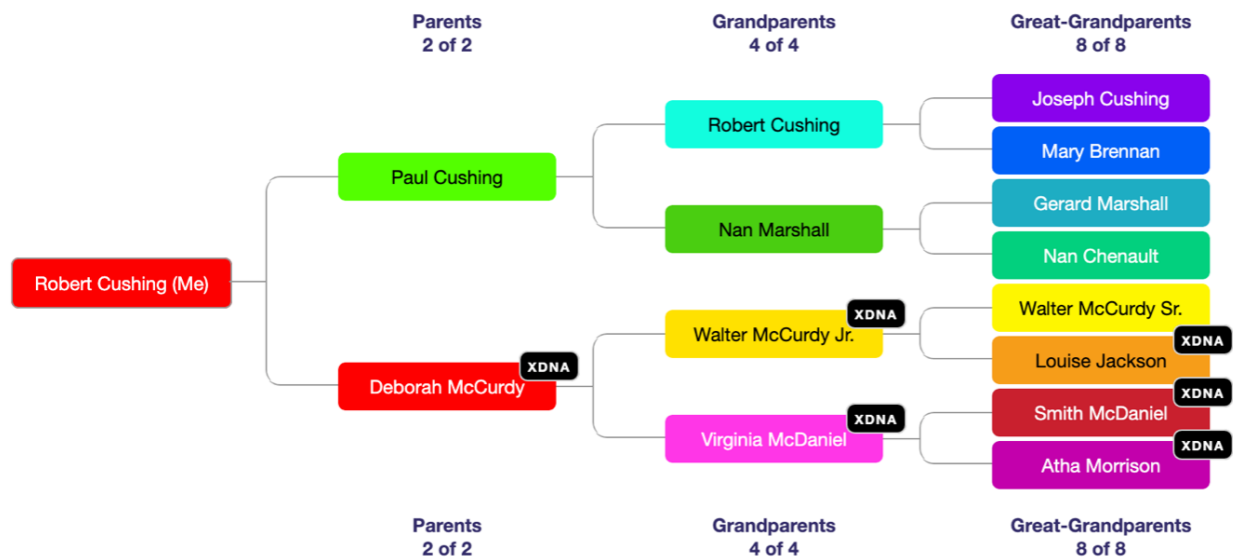
HOW IS X-DNA INHERITED?

- Men have one X chromosome that they inherit from their mothers. A man's single X is usually a blend of his mother's two X chromosomes. Men inherit Y chromosomes from their fathers meaning no X is passed from father to son.
- Women have two X chromosomes, one from each parent. A woman's maternal X is usually a blend of her mother's two X chromosomes. Her paternal X will be an exact copy of her father's single X chromosome.
- **Recombination** is when a pair of chromosomes is blended together to form a single new chromosome to be passed to a child. When a woman has a child, her two X chromosomes are typically **recombined** into a single new X. This new X will have portions of the mother's paternal and maternal X chromosomes, but some genetic data will be lost during recombination.

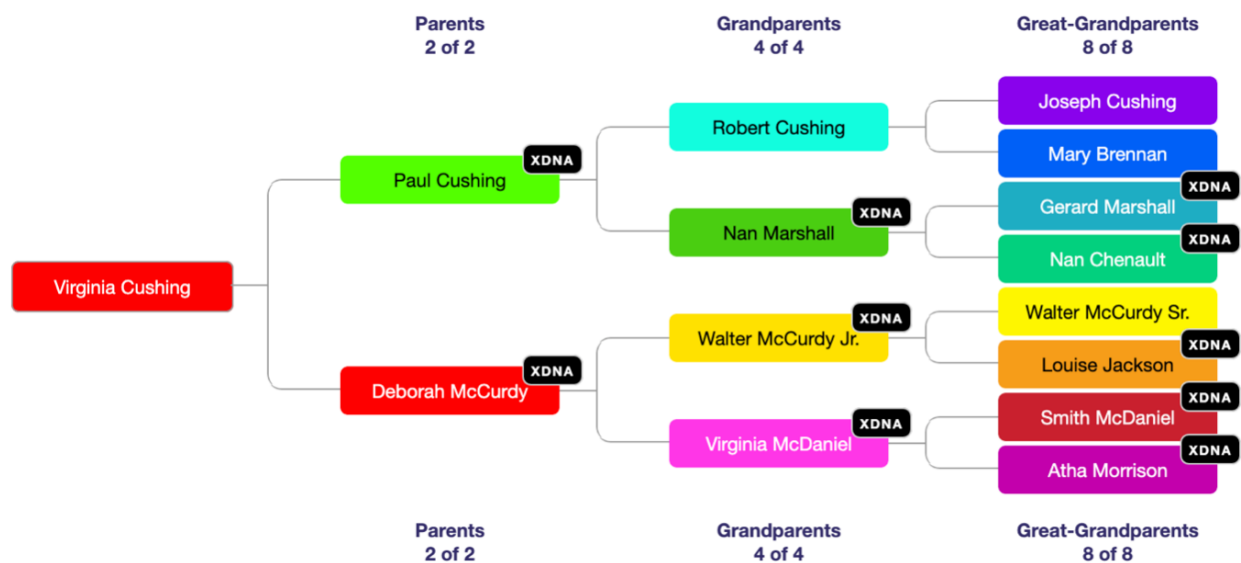


- Because recombination requires two X chromosomes, recombination can only occur when women pass X chromosomes onto their children. Since men only have one X, there is no other X chromosome with which the man's single X can recombine.
- Therefore, the maternal and paternal X chromosomes have idiosyncrasies. This is advantageous for genetic genealogists!

- In summary:
 - When a **mother** has a **daughter**, the daughter inherits a blend of her mother's two X chromosomes.
 - When a **mother** has a **son**, the son inherits a blend of her mother's two X chromosomes.
 - When a **father** has a **daughter**, the daughter inherits her father's X chromosome unchanged.
 - When a **father** has a **son**, there is no X-DNA passed down.
- Fully Identical Region: portion of chromosome where two individuals share DNA in the same location on both copies of that chromosome (*paternal and maternal copy*)
- Half Identical Region: portion of chromosome where two individuals share DNA in same location on a single copy of that chromosome (*paternal or maternal copy*)
- Full siblings will generally have a mix of fully and half identical X regions
- Half-sisters who share a father will have identical paternal X chromosomes



Typical Male X Inheritance Pattern



Typical Female X Inheritance Pattern

HELPFUL TOOLS

- DNA Painter (www.dnainter.com)
 - Offers several visualization tools for your DNA matches, notably chromosome “painting” of the X chromosome and family tree charts showing X inheritance patterns
 - Website offers thorough tutorials and FAQs

HOW TO FIND YOUR X-DNA MATCHES

	Sort by X Matches?	Segment Data?	Chromosome Browser?	Transfer to GEDmatch?
AncestryDNA	No	No	No	Yes
FamilyTreeDNA	Yes	Yes	Yes	Yes
23andMe	No	Yes	Yes	Yes
MyHeritage	No	Yes	Yes* (cannot view X)	Yes
GEDmatch	Yes	Yes	Yes	N/A

FAMILYTREEDNA FAMILY FINDER (www.familytreedna.com)

- After logging in, under “Autosomal DNA Results & Tools” click the “Family Finder Matches” button
- On your match list, look in the far right column for “X Match” (see picture below)

Haplogroup	Relationship Range	Shared DNA	Longest Block	X Match
Y-DNA: N/A mtDNA: N/A	Parent/Child SON	3564 cM	284 cM	181 cM
Match date: May 07 2021				
Haplogroup	Relationship Range	Shared DNA	Longest Block	X Match
Y-DNA: N/A mtDNA: N/A	Parent/Child SON	3530 cM	281 cM	181 cM
Match date: May 11 2021				

- Click the check box on the left of the match name of interest and then the orange “compare relationship” button that will appear in the bottom right
- This will take you to the chromosome browser where you can view relevant segment data

23ANDME (www.23andme.com)

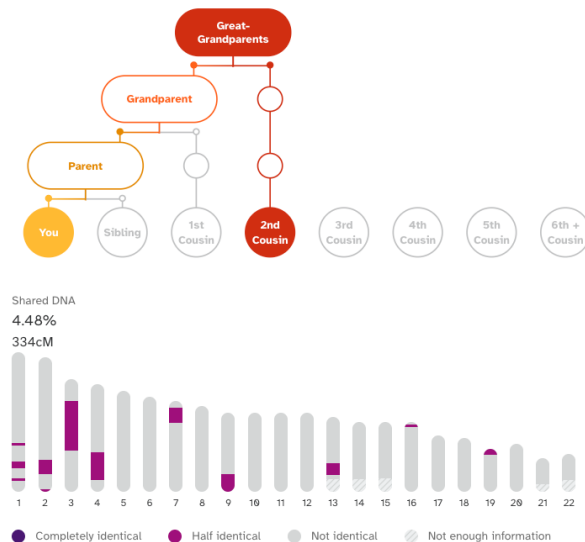
- After logging in, hover over “Ancestry & Traits” in the top menu bar and select DNA Relatives in the dropdown menu
- Click on the name of the match of interest, then on their profile page look for section “Your genetic relationship”
- On this panel, click “View DNA details” to see a visualization of your shared segments

Your genetic relationship ⓘ

Predicted relationship

2nd Cousin ↗

You and [REDACTED] may share a set of great-grandparents. You could also be from different generations (removed cousins) or share only one ancestor (half cousins).



- To download your segment data for import into DNAPainter, scroll to the bottom of the “DNA Relatives” page and click “Download DNA Relatives Data,” this will generate a .csv file that can be imported into DNAPainter

GEDMATCH

- GEDmatch offers the most robust set of X DNA analytical tools

- “One-to-One X-DNA Comparison” tool shows detailed match data on X chromosomes for two testers
- After logging on to GEDmatch (I used the Classic version), look in the right column for “DNA Applications”
- Click “One-to-One X-DNA Comparison”
- Enter kit numbers and click the “compare” button at the bottom
- (For X chromosome comparisons, I opt to select “prevent hard breaks” at the bottom of the form – if left unchecked, GEDmatch will leave out certain areas of the chromosome where the read may be less certain such as centromeres)
- Result will look something like this if there is an X match:

Chr	B37 Start Pos'n	B37 End Pos'n	Centimorgans (cM)	SNPs
23	2,700,157	52,067,217	82.7	5,729
23	61,992,011	154,892,230	105.1	8,188

Chr 23



- Blue bar indicates area of matching segments (in this instance, a match between father and daughter)
- The green lines above the blue technically indicate fully identical regions, meaning the DNA sequence is identical on both sides of the X chromosome, but unless there is a substantially lengthy portion of solid green it is almost certainly a coincidence)
- The yellow lines are half identical, meaning there is only a match to one side of the X

CASE STUDY #1: MAXINE

- Maxine was adopted in 1950 in Virginia
- Close DNA match on maternal side led to identification of Maxine’s mother
- Majority of remaining paternal matches were distant
- Closest paternal match was Samuel Miller, sharing 433 cM of autosomal DNA and 100 cM of X DNA
- Evaluated all possible X ancestral lines for most likely connection
- Use significant X matches to make your research more efficient
- Moral of the story: Start with relationship hypothesis with the fewest possible recombination events (fewer opportunities for X DNA to be diluted and “lost”)

CASE STUDY #2: SHIRLEY

- Shirley was born in 1935 in Chicago, no knowledge of father
- Based on autosomal results, narrowed her placement in larger family tree to three likely positions
- What Are the Odds tool (www.dnainter.com/tools/wato) indicates the three hypotheses are equally likely
- When X DNA is considered we can rule out 2/3 of the hypotheses, allowing us to focus our research efforts towards finding additional evidence on the most likely hypothesis

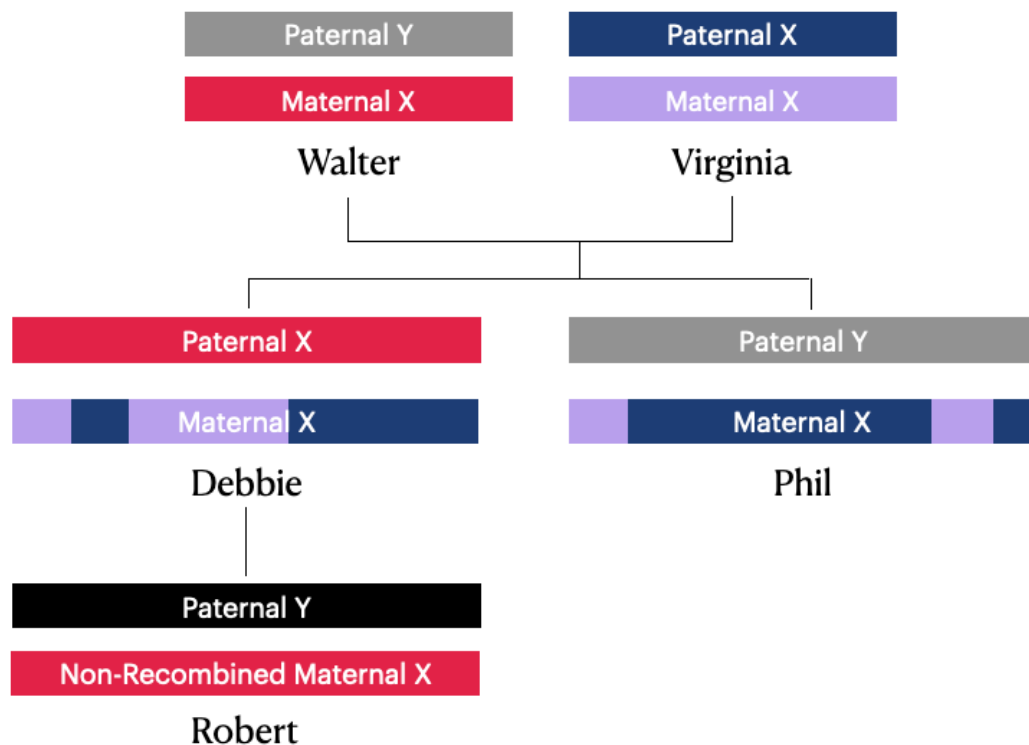
- Moral of the story: When autosomal DNA isn't providing any compelling leads, check for shared X DNA, let X DNA guide your research when applicable

CASE STUDY #3: MARIE

- My mother and Marie share 90 cM of autosomal DNA and 26 cM of X DNA
- They are confirmed 2C2R through common ancestors Jesse Haralson and Mary Dacus
- However, that connection contains two instances of male-male generational linkages, ruling out the possibility of shared X DNA through the Haralson line
- Further research determines that my mother and Marie share a second set of common ancestors on a more distant X line
- Moral of the story: always double check that an X match makes sense with your suspected common ancestors, if not research

X-CEPTION TO THE RULE: NON-RECOMBINANT X CHROMOSOMES

- When a woman passes one of her X chromosomes unchanged to her child, this is called a **non-recombination event**
- In other words, a child will inherit one of their mother's two X chromosomes rather than a blend of the two
- This diagram shows an instance from my own family tree: I inherited my mother's paternal X chromosome without recombination, meaning I share no X DNA with my maternal grandmother



IN SUMMARY

- X DNA is a key tool for genetic genealogists
- By understanding its unique inheritance pattern, we can strengthen our genealogical conclusions
- Like all DNA evidence, it must be correlated with other evidence in order to meet the Genealogical Proof Standard

SUGGESTED FURTHER READING

Bettinger, Blaine T. “X Chromosomal (X-DNA) Testing.” In *The Family Tree Guide to DNA Testing and Genetic Genealogy* (2nd Edition). Family Tree Books, 2019.

Bettinger, Blaine T. and Debbie Parker Wayne. “Genealogical Applications for X-DNA.” In *Genetic Genealogy in Practice*. National Genealogical Society: Arlington, Virginia, 2016.

Brons, Mercedes. “What is an X DNA Match on Family Tree DNA?” *Who Are You Made Of?* (<https://whoareyoumadeof.com/blog/what-is-an-x-dna-match-on-family-tree-dna/> : accessed 8 February 2022).

Estes, Roberta. “That Unruly X... Chromosome That Is,” *DNAeXplained – Genetic Genealogy*, (<https://dna-explained.com/2014/01/23/that-unruly-x-chromosome-that-is/> : accessed 8 February 2022).

Johnston, Kathryn J., MD. “X-DNA Techniques and Limitations.” In *Advanced Genetic Genealogy: Techniques and Case Studies*, ed. Debbie Parker Wayne. Cushing, Texas: Wayne Research, 2019.

Smith, Jared. “X Chromosome Recombination’s Impact on DNA Genealogy.” (<https://smithplanet.com/stuff/x-chromosome.htm> : accessed 8 February 2022).